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The National Center on Birth Defects and Developmental Disabilities:

Past, Present, and Future

Coleen A. Boyle, PhD, MS hyg, José F. Cordero, MD, MPH, and Edwin Trevathan, MD, MPH
National Center on Birth Defects and Developmental Disabilities (Boyle), CDC, Atlanta, Georgia; the School of Public Health (Trevathan), St. Louis University, St. Louis, Missouri; and Graduate School of Public Health (Cordero), University of Puerto Rico, San Juan, Puerto Rico

The National Center on Birth Defects and Developmental Disabilities (NCBDDD) was established in April 2001 as a result of *The Children's Health Act of 2000*.¹ The supporters of the center's creation wanted to raise the visibility of child health and disability at the CDC. From its inception, the NCBDDD included the established programs in birth defects, genetic disorders, developmental disabilities and disabilities and health, with a more than 30-year history in surveillance, research, and public health practice, including the training of public health professionals in these respective fields.

In 2003, the NCBDDD expanded with the addition of the blood disorders program, which initially had been established in response to the discovery that HIV, a bloodborne infection, highly affected people with hemophilia who required treatments using blood products. This led to the development of a comprehensive program to ensure optimal care for this and other populations affected by nonmalignant blood disorders.² The current paper examines the health impact of the center's work that was highlighted in a year-long reflection, "10 Years of Service"³; and provides a framework for how the NCBDDD will move forward with a renewed emphasis on enhancing public health and healthcare-system capacity to have greater health impact for the populations served.

10 Years of Service: 10 Years of Making a Difference

In 2001, guided by earlier NCBDDD scientific findings, the center published the first evaluation showing that folic acid fortification of enriched cereal grain products reduced the prevalence of neural tube defects (NTDs) in the U.S. by 31%.⁴ As a result of the implementation of mandatory folic acid fortification in the U.S., the NCBDDD estimates that each year, 1000 more babies are born without an NTD.⁵ Given the progress in preventing NTDs in the U.S.,⁵ the NCBDDD has turned its attention to the global elimination of folic acid-preventable NTDs with initial efforts in Southeast Asia where the prevalence of this condition is highest.⁶

Address correspondence to: Coleen A. Boyle, PhD, MS hyg, National Center on Birth Defects and Developmental Disabilities, CDC, 1600 Clifton Rd, Atlanta GA 30333. cboyle@cdc.gov.

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Being vigilant and responding quickly to health threats is a critical activity for the NCBDDD. As an example, in 2002, in collaboration with the Food and Drug Administration, scientists at the center found that children with a cochlear implant were much more likely to develop bacterial meningitis than children in the general population.⁷ As a consequence, the cochlear device associated with the highest risk of meningitis was withdrawn from the market, clinical treatment guidelines were revised for new and current device recipients, and national vaccine recommendations were strengthened so that cochlear implant recipients would be considered a high-risk group during times of vaccine shortage.⁸

Deep vein thrombosis (DVT) is a common and serious blood disorder. In 2003, under the guidance of the NCBDDD, more than 60 organizations assembled at the Public Health Leadership Conference on DVT to discuss the urgent need to make this condition a major U.S. public health priority. In 2008, the NCBDDD supported the Surgeon General's "Call to Action to Prevent Deep Vein Thrombosis (DVT) and Pulmonary Embolism," which highlighted the public health need for prevention, with more than 600,000 people developing a DVT and 100,000 deaths each year.⁹ Prevention of DVT, particularly that associated with hospitalization, has become an NCBDDD priority. The work will focus on system-level strategies to improve uptake of hospital protocols for DVT prevention and the development of a monitoring system to track compliance and the impact of these measures on DVT prevention.¹⁰

In 2004, the NCBDDD, working with many partners including American Academy of Pediatrics, Autism Speaks and the Autism Society, launched the "Learn the Signs. Act Early." campaign. This effort was focused on the importance of monitoring a child's development, and increasing knowledge of early signs of autism and other developmental disabilities; emphasis was placed on encouraging parents and healthcare providers to act early if a delay in development is suspected. An evaluation of the campaign showed an influence on provider and parent knowledge about these important actions.¹¹ The campaign also triggered educational efforts to increase early diagnosis and intervention to mitigate the challenging behaviors often associated with autism. The work of the NCBDDD in this area has now expanded to support state and local efforts to integrate the complex health and education systems involved in early screening, diagnosis, and referral for services for children with developmental delays.

The NCBDDD was a leader in the national efforts to identify disability as a major public health issue. This effort included the 2005 Surgeon General's "Call to Action to Improve the Health and Wellness of Persons with Disabilities" and the 2007 IOM's report, "The Future of Disability in America."^{12,13} These reports resulted in an appeal to all Americans to help improve the quality of life for people with disabilities through better health care and understanding. Integration of the needs of people with disabilities into mainstream health promotion programs, improving healthcare access, and ensuring that emergency preparedness programs reach people with a range of functional disabilities are key priorities for the NCBDDD. The center is working to align funded state public health programs and grants to consumer and support groups with the three priorities. Promoting disability inclusion into the CDC's mainstream public health surveys and programs is also a top priority.

In 2006, the NCBDDD and its partners initiated a national campaign to promote and improve the preconception health of women, couples, and families. As a result, the first national preconception conference was convened, and national guidelines for improving preconception health and health care were published as part of an *MMWR Reports and Recommendations*.¹⁴ This seminal work has reframed the focus of healthy mothers and healthy pregnancies from the prenatal period to one that encompasses pregnancy planning and preconception health as critical for healthy babies. Efforts have resulted in an IOM report recommending preconception health as an essential women's health service.¹⁵ Similarly, the WHO is undergoing a revision of their maternal, infant, and child health life span framework to include a preconception health component.

In 2007, through the Autism and Developmental Disabilities Monitoring Network, the NCBDDD issued its first report on the prevalence of autism spectrum disorders (ASDs), indicating an average of one in 152 children aged 8 years identified with autism.¹⁶ This was followed by reports in 2009 and 2012 updating the prevalence estimate to an average of one in 88 children with an ASD.^{17,18} The NCBDDD will continue to work with state health departments and other partners to monitor trends and prioritize resources and activities in order to understand why the prevalence is changing.

Also over this time frame, the NCBDDD-supported National Birth Defects Prevention Study, conducted in nine sites in the U.S., showed the important role of a number of modifiable maternal risk factors (e.g., preexisting chronic medical conditions) in the risk of birth defects. Among these risk factors was maternal prescription medication use (e.g., selective serotonin-reuptake inhibitors, opioid analgesics, and asthma medications).^{19–21} This growing body of evidence highlights the need for accurate data to inform women and their healthcare providers about safe use of medications in pregnancy and sparked the NCBDDD “TR_x eating for Two” initiative.

Finally, based on the results of an NCBDDD-sponsored clinical trial of regular presymptomatic infusions with Recombinant Factor VIII, this preventive treatment has become the standard of care in the U.S. for people with hemophilia, subsequently leading to important reductions in the joint pain and other complications pervasive in this disorder.²² The NCBDDD is updating its comprehensive surveillance program for hemophilia to identify other health-promoting treatment strategies, particularly as the population of people with hemophilia ages.

Data for action are at the core of the NCBDDD mission. Two recent projects are important examples of how data are provided to positively influence population health. First, in 2009, the NCBDDD, in collaboration with the NIH's National Heart Lung and Blood Institute, developed a model surveillance system for sickle cell disease (SCD) and thalassemia called the Registry and Surveillance for Hemoglobinopathies (RuSH), the first-ever system of its kind. This effort will provide much-needed data to understand gaps in healthcare delivery of life-prolonging therapies for SCD. Second, to identify system-level opportunities for the NCBDDD's priorities on disability, in 2011, the NCBDDD released a web-based state-specific data system, the Disability and Health Data System, which aggregates key risk factor and other disparity data from CDC's Behavioral Risk Factor Surveillance System by

disability status. This electronic application will provide ongoing information to guide health promotion and healthcare policy decisions at the federal and state levels.

Partnerships for Health Promotion

Much of the work of the NCBDDD during the past 10 years would not have been possible without the support of key partners and constituents. A series of field events and activities were held in Atlanta GA, Denver CO, Trenton NJ, Nashville TN, and Washington DC to showcase the important work of our collective organizations. Each event highlighted an NCBDDD priority and featured people in the local community who helped to champion the work of the center.

Building the Future

To commence the second decade, the NCBDDD developed a 5-year strategic plan. The plan was guided by a series of formal peer-reviewed program evaluations conducted in 2006–2007, coupled with an internal process of prioritization that relied on a standard set of public health criteria. Emphasis was placed on those activities that could have demonstrable health impact in the next 3–5 years. A draft of the plan was vetted with stakeholders representing the center's various populations. The foundation of the plan was to identify and commit to accomplishing activities in six focus areas. Although the selected focus areas did not indicate a lessening of commitment to other important work, they were highlighted in the plan to ensure alignment of the NCBDDD's strengths and resources with critical needs and existing opportunities in an effort to positively affect relevant populations under its purview.

The six focus areas are to:

- prevent major birth defects attributable to maternal risk factors, with an initial focus on the global elimination of folic acid-sensitive NTDs and the prevention of birth defects associated with teratogenic exposures including prescription and over-the-counter medications and alcohol;
- enhance tracking, research, and awareness of ASDs to monitor changes in occurrence, identify preventable risk factors, and facilitate early diagnosis;
- prevent death and disability associated with DVT/pulmonary embolism, with a focus on such events associated with hospitalization;
- prevent and control complications of SCD through improved uptake of preventive treatments;
- reduce disparities in obesity and other health indicators in children, youth, and adults with disabilities through general and targeted health promotion;
- promote inclusion of disability into mainstream public health, with a focus on the use of disability identifiers in CDC surveys and surveillance systems and integrating disability into key CDC public health promotion programs and research.

Non Satis Scire: “To Know Is Not Enough”

In addition to the traditional public health approaches that engage research, surveillance, and policy, the NCBDDD’s future focus will engage system-level strategies to positively influence health by closing the gap between existing public health, clinical, and biomedical interventions and their application, and full implementation on a population level. For instance, it is estimated that 40% of deaths from DVT/pulmonary embolism could be prevented if guidelines for appropriate prophylaxis were fully implemented within the hospital setting.²³ The NCBDDD will work with other federal agencies, hospital associations, and clinical professional groups to devise a strategy to affect uptake and effective use of the evidence-guided recommendations.

Similarly, data are accumulating for clinical decision making about the safe use of specific medications in pregnancy. However, this information is not uniformly evaluated and readily available or easily interpreted by healthcare providers and consumers. The NCBDDD will take a leadership role in engaging other invested federal agencies (e.g., Food and Drug Administration and the NIH) and professional organizations (e.g., American Congress of Obstetricians and Gynecologists) to develop an ongoing systematic review process to evaluate the accumulating evidence on maternal and fetal risk associated with commonly used prescription medications and develop a health information framework for its dissemination to primary healthcare providers and consumers.

Conclusion

The 10 years since the creation of the NCBDDD has seen important changes in the health of the populations that are included in the center’s mandate. In moving forward, the NCBDDD has identified a number of achievable priorities that will guide its work over the next decade. The recognition that the greatest impact occurs through policy-and systems-level applications will influence how the work gets done. Although the center is only one player among many, it is emboldened to take a leadership role as convener in moving these priorities forward.

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